Neuromuscul Disord. 2017 Dec 13. pii: S0960-8966(17)31365-2. doi: 10.1016/j.nmd.2017.12.005. [Epub ahead of print]

DOK7 myasthenic syndrome with subacute adult onset during pregnancy and partial response to fluoxetine.

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Abstract

DOK7 congenital myasthenic syndrome (DOK7-CMS) generally presents early in life and is treated with salbutamol or ephedrine. This report describes an atypical case of a 39-year-old woman who presented with proximal upper limb weakness in the third trimester of pregnancy and was initially diagnosed with seronegative myasthenia gravis. Dramatic clinical worsening under pyridostigmine and further inefficacy of steroids, intravenous human immunoglobulin (IVIG) and plasma exchange (PLEX) led to the presumptive diagnosis of a CMS. Initially, a slow-channel CMS was regarded as more probable due to prominent finger extension weakness. Accordingly, fluoxetine was started and a lengthy improvement was seen. Clinical deterioration occurred after fluoxetine withdrawal, when a c.1124_1127dup homozygous mutation was detected in DOK7 gene. Afterwards, salbutamol was started and the patient became asymptomatic. This case highlights the importance of considering CMS before an adult-onset myasthenic syndrome and suggests a benefit from fluoxetine not previously reported in DOK7-CMS.

KEYWORDS: Congenital myasthenic syndrome; DOK7; Fluoxetine; Pregnancy

PMID: 29395672 DOI: 10.1016/j.nmd.2017.12.005

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